

(12) INTERNATIONAL PUBLICATION PUBLISHED UNDER THE PATENT COOPERATION TREATY (PCT)

(19) World Intellectual Property
 Organization
 International Bureau



(43) International Publication Date
 15 July 2004 (15.07.2004)

PCT

(10) International Publication Number
WO 2004/058985 A2

(51) International Patent Classification⁷:

C12Q

(74) Agents: **HODGES, Robert, A.** et al.; Needle & Rosenberg, P.C., 999 Peachtree Street, Suite 1000, Atlanta, GA 30309-3915 (US).

(21) International Application Number:

PCT/US2003/040278

(22) International Filing Date:

17 December 2003 (17.12.2003)

(25) Filing Language:

English

(26) Publication Language:

English

(30) Priority Data:

60/433,774

17 December 2002 (17.12.2002)

US

(81) Designated States (*national*): AE, AG, AL, AM, AT, AU, AZ, BA, BB, BG, BR, BW, BY, BZ, CA, CH, CN, CO, CR, CU, CZ, DE, DK, DM, DZ, EC, EE, EG, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MA, MD, MG, MK, MN, MW, MX, MZ, NI, NO, NZ, OM, PG, PH, PL, PT, RO, RU, SC, SD, SE, SG, SK, SL, SY, TJ, TM, TN, TR, TT, TZ, UA, UG, US, UZ, VC, VN, YU, ZA, ZM, ZW.

(84) Designated States (*regional*): ARIPO patent (BW, GH, GM, KE, LS, MW, MZ, SD, SL, SZ, TZ, UG, ZM, ZW), Eurasian patent (AM, AZ, BY, KG, KZ, MD, RU, TJ, TM), European patent (AT, BE, BG, CH, CY, CZ, DE, DK, EE, ES, FI, FR, GB, GR, HU, IE, IT, LU, MC, NL, PT, RO, SE, SI, SK, TR), OAPI patent (BF, BJ, CF, CG, CI, CM, GA, GN, GQ, GW, ML, MR, NE, SN, TD, TG).

(71) Applicant (*for all designated States except US*): **UNIVERSITY OF UTAH RESEARCH FOUNDATION** [US/US]; 615 Arapen Drive, Suite 110, Salt Lake City, UT 84108 (US).

(72) Inventors; and

(75) Inventors/Applicants (*for US only*): **FLANIGAN, Kevin, M.** [US/US]; 131 J Street, Salt Lake City, UT 84103 (US). **WEISS, Robert, B.** [US/US]; 4273 South 3080 East, Salt Lake City, UT 84124 (US). **DUNN, Diane, M.** [US/US]; 3382 E. Larchmont Dr., Salt Lake City, UT 84109 (US). **NIEDERHAUSERN, Andrew von** [US/US]; 449 Wendell Way, Salt Lake City, UT 84115 (US).

Published:

— without international search report and to be republished upon receipt of that report

For two-letter codes and other abbreviations, refer to the "Guidance Notes on Codes and Abbreviations" appearing at the beginning of each regular issue of the PCT Gazette.

(54) Title: **RAPID DIRECT SEQUENCE ANALYSIS OF MULTI-EXON GENES**

(57) Abstract: Disclosed is a Single Condition Amplification/Internal Primer (SCAIP) sequencing method which allows for the rapid, accurate, and economical analysis of any large multi-exon gene. The method can be used to detect genomic mutations in any large multi-exon gene including the dystrophin gene. In some forms, the method can rely on amplification of a large number of exons at a single set of PCR temperatures with a first set of amplification primers followed by sequencing without optimization of individual amplicon conditions, using a second, internal set of sequencing primers. The SCAIP method provides for the identification and analysis of specific individual genomic mutations such as deletions, point mutations, frameshifts, or combinations thereof, in gene complexes with multiple exons/introns spanning large genomic regions.



WO 2004/058985 A2